CTNND1 gene

catenin delta 1

Normal Function

The *CTNND1* gene provides instructions for making a protein called p120-catenin, also known as delta 1 catenin. This protein and others in the catenin family interact with proteins from another family called cadherins. Catenins and cadherins form protein complexes that help cells stick to neighboring cells (cell adhesion) to form organized tissues. In addition to their role in cell adhesion, catenin and cadherin interactions help transmit chemical signals within cells, control cell maturation and movement, and regulate the activity of certain genes.

The p120-catenin protein interacts with a protein called E-cadherin, which is produced from the *CDH1* gene. The E-cadherin protein is found within the membrane that surrounds epithelial cells, which are the cells that line the surfaces and cavities of the body, including the inside of the eyelids and the mouth. The p120-catenin protein helps keep E-cadherin in its proper place in the cell membrane, preventing it from being taken into the cell through a process called endocytosis and broken down prematurely. Interactions between the two proteins are also important for other cell processes that are involved in the development of the head and face (craniofacial development), including the eyelids and teeth.

Health Conditions Related to Genetic Changes

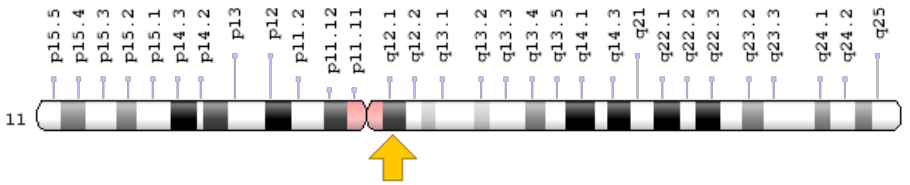
Blepharocheilodontic syndrome

At least three *CTNND1* gene mutations have been identified in people with blepharocheilodontic (BCD) syndrome, a disorder that is present at birth and causes abnormalities mainly affecting the eyelids and mouth. These mutations are thought to prevent the production of the p120-catenin protein or result in an unstable protein that quickly breaks down. A shortage of p120-catenin protein allows E-cadherin to be taken into the cell and destroyed too quickly by endocytosis. A shortage of these proteins disrupts normal development, especially craniofacial development, which is thought to underlie the features of BCD syndrome.

Chromosomal Location

Cytogenetic Location: 11q12.1, which is the long (q) arm of chromosome 11 at position 12.1

Molecular Location: base pairs 57,761,802 to 57,819,540 on chromosome 11 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- cadherin-associated Src substrate
- CAS
- catenin (cadherin-associated protein), delta 1
- CTNND
- delta catenin
- KIAA0384
- p120
- p120 catenin
- p120(CAS)
- p120(CTN)
- P120CAS
- P120CTN

Additional Information & Resources

Educational Resources

- Developmental Biology (sixth edition, 2000): Morphogenesis and Cell Adhesion https://www.ncbi.nlm.nih.gov/books/NBK10021/

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28CTNND1%5BTIAB%5D%29+OR+%28catenin+delta+1%5BTIAB%5D%29%29+OR+%28%28CTNND%5BTIAB%5D%29+OR+%28P120CAS%5BTIAB%5D%29+OR+%28P120CTN%5BTIAB%5D%29+OR+%28cadherin-associated+Src+substrate%5BTIAB%5D%29+OR+%28catenin++,+delta+1%5BTIAB%5D%29+OR+%28p120+catenin%5BTIAB%5D%29+OR+%28p120%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- CATENIN, DELTA-1
  http://omim.org/entry/601045

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/CTNND1ID40197ch11q11.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=CTNND1%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:2515
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:1500
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/1500
- UniProt
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